Flecked-retina Disorders: Deletion/Duplication Panel

**Test Code:** MD134

**Turnaround time:** 2 weeks

**CPT Codes:** 81228 x1, 81479 x1

**Condition Description**

Flecked-retina refers to a varying distribution and size of yellowish lesions within the retina (optic nerve or vascular anomalies are not generally observed). Flecked-retina disorders may be stationary or progressive and range from benign to visually debilitating. The term “flecked-retina” is generally not favored by retina specialists due to the distinctive pattern and clinical course of each individual disorder. Examples of conditions in this category include benign flecked-retina, fundus flavimaculatus (Stargardt's), and fundus albipunctatus.

**References:**
- OMIM
- GeneReviews
- Emory and Rimoin's Principles and Practice of Medical Genetics, 5th Edition

**Genes**

ABCA4, PLA2G5, PRPH2, RDH5, RHO, RLBP1

**Indications**

This test is indicated for:
- Confirmation of a clinical diagnosis of flecked-retina disorders.
- Carrier testing in adults with a family history of flecked-retina disorders.

**Methodology**

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region. Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

**Detection**

**Deletion/Duplication:** Detection is limited to duplications and deletions. The CGH array will not detect point or intronic pathogenic variants. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

**Specimen Requirements**

Submit only 1 of the following specimen types

**Type: Whole Blood**

Specimen Requirements:
- In EDTA (purple top) tube:
  - Infants (2 years): 3-5 ml
  - Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Specimen Requirements:
- In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.
Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

**Special Instructions**

Please include fundus photographs, electroretinogram (ERG) findings, visual field findings, and visual acuity, if available, for expert review and clinical correlation with test results.

**Related Tests**

- Eye Disorders: Comprehensive Sequencing and Deletion/Duplication Panels.
- Flecked-retina Disorders: Sequencing Panel.